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Introductory Chapter: Dystonia - Different Prospects

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1. Introduction

Movement disorders are considered one of the most controversial diseases when it comes to diagnosis, classification, and evidence-based lines of management, each case is different, and it can be quite challenging to get these movements under control [1].

Dystonia is a wide-spectrum disorder which affects all age groups. Etiology of dystonia is usually diverse; in some cases within the pediatric age group, the presence of spasticity can make the diagnosis more difficult. For example, cerebral palsy patients can suffer from dystonia and spasticity, though most of pediatric cases with cerebral palsy will suffer from spasticity alone. Dystonia may result from either diffuse or localized pathology of the cerebral cortex, brain stem, or spinal cord. Management of dystonia is challenging, and specific goals should be identified. Dystonia is considered one of the most disabling conditions in pediatric age group which may remain till adulthood, treatment is usually unsatisfactory, and patients will show limited response to pharmacotherapy [1].

Dystonia is a neurological disorder of patterned, involuntary, repetitive, or sustained contraction of antagonist group of muscles. This results in twisting movements and ends in abnormal posturing. Primary dystonia is considered the most common form of dystonia encountered through neurology practice. Isolated dystonia usually starts during childhood years or early adolescence. It usually runs a progressive course with marked disability and functional impairment. Recent advances in genetics elaborated more than the well-known monogenic forms of isolated dystonia due to DYT gene defects; the etiological spectrum is now widened with great variance in phenotype. Many genes which were thought to be responsible to cause certain disease were recently proven to be responsible for dystonia as well which results in more complex phenotype [2].

Current treatment aims to help decrease the frequency of abnormal involuntary movements, improve posturing, and prevent the subsequent development of contractures. This will reduce

pain and facilitate better quality of life. These include oral pharmacotherapy, e.g., trihexyphenidate (Level A Class I–II); botulinum toxin injection for cases of localized or segmental dystonia, e.g., cervical dystonia (Level A Class I–II); and surgical interventions like intrathecal baclofen pump; and deep brain stimulation (DBS) for cases with generalized dystonia who failed oral pharmacotherapies [3].

2. Management modalities

Intrathecal baclofen proved to be effective in progressive dystonia in selected cases. Deep brain stimulation is more effective in controlling primary generalized dystonia. Intracranial stimulation of the postero-latero-ventral region of the internal globus pallidus in patients with dystonia-choreoathetosis cerebral palsy significantly improved their level of functionality and motor abilities, reduces their pain, and improved their overall quality of life. Synergetic effect was proved to be highly effective in treating primary generalized dystonia where both surgical modalities were used and produced excellent control of dystonia. It can reach up to 97% improvement regarding hygiene, posturing, and daily life activities [3].

Medical cannabis have been explored in a variety of medical conditions which are not controlled with the current treatment modalities. Cannabidiol oil has been studied in pediatric cases with complex motor disorders; significant improvement in control of spasticity and dystonia was achieved; it was well tolerated and showed good effect on sleep pattern, pain, and overall quality of life [4].

3. Presentations

Dystonia has various forms of presentations; one of the popular forms is Meige's syndrome or "oromandibular dystonia"; it is considered as one of the most important forms of focal dystonia as it may be misdiagnosed as temporomandibular joint disorder or psychogenic disorder which will alter the management plan and delay proper treatment. That chapter describing oromandibular dystonia will cover its etiology, diagnosis, and available lines of management. Furthermore, the consensus update will be discussed, with recent classification of dystonia focusing on its clinical characteristics and etiology. The first focuses on the age of onset, body distribution, chronological pattern, coexistence of other movement disorders, and other neurological manifestations. The body distribution is classified to focal, segmental, multifocal, generalized, or hemi-dystonia. Associated features distinguish isolated dystonia in both genetic and idiopathic cases that are often resembled to as primary dystonia and combined dystonia. The second axis allows further division according to the presumed etiology.

Although dystonia is a rare condition in general population, the "pure," primary, and isolated dystonia is the third most common movement disorder, after essential tremor and Parkinson's disease; dystonia and associated disorders influenced a major impact on quality of life.

Concepts on phenomenology have also renewed with decades, considering dystonia to be a solely motor disorder to an increasing recognition of associated neurological or psychiatric features which indicate that the disorder is not purely motor. This resembles the growing knowledge on dystonia's pathophysiology where the recent insights from neurophysiologic studies identified functional abnormalities in the basal ganglia sensorimotor network and, more recently, the cerebellothalamocortical pathway. Besides the well-known lack of inhibition at different nervous system levels, dystonia is specifically characterized by abnormal sensory feedback, maladaptive plasticity in the sensorimotor cortex, and loss of cortical surround inhibition.

Bearing in mind the new classification, where the term "primary" is no more recommended, and the circumstance that in majority of cases it resembles the new and more precise term "isolated," both terms are used in this chapter, in order to be correct when providing information from the studies cited, fully understanding that some time and efforts are needed to replace completely the old terminology with the new one.

Dystonia with non-motor co-morbidities, including sleep and psychiatric disorders, cognition, as though as pain and sensory symptoms, their pathophysiological and biochemical mechanisms, relations with the symptomatic treating strategies for the abnormal movements, and specific treatment for the non-motor signs.

Many antipsychotic drugs have been used to control these comorbid conditions. Neuroleptics caused variety of intolerable side effects, among which drug-induced movement disorders could be seriously problematic. Tardive syndromes, represented by tardive dyskinesia and tardive dystonia, still remain challenging in many respects.

"Dystonia tarda" was a term firstly used in 1973 to define a dystonia which appeared as a delayed undesirable effect in patients exposed to neuroleptic drugs. Case descriptions appear in the literature even earlier, but a definition of the term "tardive dystonia" came from Burke et al. [5] who also implemented criteria for the diagnosis. The syndrome consists of involuntary, sustained muscle contractions, usually slow, often painful, affecting the face, neck, limbs, or trunk. The involuntary muscle contraction(s) often cause abnormal postures and twisting movements, which are often disfiguring and socially awkward.

Tardive syndromes manifest in a variety of persistent motor and sensory syndromes, manifesting as an adverse reaction from antidopaminergic agents or, less often, by other types of drugs. Tardive dyskinesia was the first to be observed, in the 1950s, typically comprising rhythmic, repetitive oro-buccal-lingual involuntary movements, which however can also appear in the trunk, limbs, and pelvis. Later, it was also described as a distinct, frequently co-occurring, or independently manifesting condition. Although the term "tardive" implicates a long exposure and delayed onset, tardive syndromes can actually show up even days after administration of the offending agent, notwithstanding that risk increases with longer exposure durations. Another important feature is their persistent nature, meaning that they persist or even worsen following discontinuation of the offending drug.

Researchers have difficulties to deal with it separately. Moreover, there is often some confusion in the literature, as authors may sometimes use the term tardive dyskinesia in order to

refer to a variety of tardive syndromes. Much work examines tardive syndromes in general, and it is difficult to extract data referring specifically to tardive dystonia. Still, the distinction and separate examination are important, because it differs from tardive dyskinesia in respects of presentation, course, prognosis, and treatment; it is frequently more debilitating and treatment resistant; it is associated with poorer quality of life, reduced treatment compliance, and psychiatric morbidity. Another task is to differentiate tardive dystonia from acute dystonia, emerging acutely and within days after the initial administration of anti-D2 agents, in young males, as well as from other types of dystonia.

In their pioneering work, Adityanjee et al. [6] had hoped that tardive dystonia together with tardive dyskinesia would in the future be a matter of historical interest, thanks to the advent of new, better antipsychotics. Twenty years later, these movement disorders are not at all lost, not yet forgotten. Continuing to be a serious burden, they call for better understanding, prompt recognition, prevention, and better treatment.

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